FINAL PUBLISHABLE SUMMARY REPORT

INTRODUCTION AND OBJECTIVES: Direct-to-consumer (DTC) genetic testing can be understood as involving the direct access or ordering of genetic tests without the intermediate of a health-care professional from the traditional health care system and/or the advertising of genetic tests directly to the public. In the two years since this project started (May 2009), the rapid increase in number of companies offering such services has sparked an ongoing debate between different stakeholders regarding the related risks and benefits. The overall goal of this research project was to study and reveal the ethical, legal and social aspects (ELSA) of DTC genetic testing. Specifically we aimed: 1) to offer a description and review of the existing companies, the way they function and the types of tests offered; 2) to address some legal aspects of such testing in North America and Europe; 3) to collect data on the awareness, experiences and attitudes of clinical geneticists regarding DTC genetic testing; 4) to discuss specific types of genetic tests sold directly-to-the public and present them as case studies in order to address a wide variety of ELSA; 5) to disseminate the project results and stimulate international collaborations on research addressing the commercialization of genetic services.

DESCRIPTION OF WORK PERFORMED: Since this project began in May 2009, we have published 14 peer reviewed articles on the subject of DTC genetic testing in international journals, including, among others, Nature Reviews Genetics, EMBO Reports, and the European Journal of Human Genetics. We have also published one book chapter[1] and one project report.[2] Two other publications are under review. We have conducted a review of existing DTC genetic testing companies[3] and described the model of provision and summarized some important aspects of the market.[4] With respect to social and ethical aspects, one of the main themes addressed is the inappropriate testing of minors within the DTC genetic testing context.[3, 5] This also included an empirical study in which we directly surveyed companies to get more information regarding their testing policies.[6] We have also addressed the issue of consent and research conducted by DTC genetic testing companies[7], as well as the problems with consumer understanding of the medical value of DTC genetic tests and the disclaimers made by companies.[8] In addition, we have surveyed clinical geneticists to obtain their views on DTC genetic testing. We also studied blogs of users to investigate their motivations to purchase these tests. We have additionally focussed on some specific types of tests sold DTC, namely, preconceptional tests[9], psychiatric tests[10] and pharmacogenomic tests.[11] Moreover, legal aspects of DTC genetic testing have been addressed in a review of European laws in seven countries (in review). With respect to our dissemination objectives we have presented our data regularly at seminars and at international academic meetings including the European Association of Human Genetics (2009, 2010, 2011), the American Association of Human Genetics (2009), HUGO conference (2009), and a Genome Canada Meeting(2010). Moreover we have organized workshops at the European Society for human Genetics meeting in 2009 and 2010. With the collaboration of Dr. Bartha Knoppers we have set up an Internet data base of articles and normative documents regarding DTC genetic testing called DTCgen (http://www.dtcgenetest.org/).

SELECTED RESULTS: Our major results include the following: 1) There is a great heterogeneity of genetic tests presently on offer by different companies mostly situated in the USA. This heterogeneity means that ethical, legal and social issues may vary with different types of tests and different companies. 2) Furthermore, some DTC companies are allowing for children under the age of majority to be tested for adult onset diseases. This clashes with professional guidelines that state that for predictive genetic testing to be conducted in asymptomatic minors there should be therapeutic or preventive measures available. 3) A number of companies selling DTC genome-wide-testing such as 23andME, deCODE and Navigenics are conducting research using consumers' data. The activities of companies offering DTC genetic testing have not only blurred the lines between medical services and consumer products, but also between these two activities and research. As a consequence, the appropriate treatment and autonomy of individuals who purchase DTC genetic testing services could be undermined. 4) We have also revealed the confusion that exists surrounding the disclaimers that DTC genetic testing companies make on their websites regarding the fact that their services are only educational and have no medical or diagnostic value. 5) Through the study of users' blogs we have

revealed that the main motivations to purchase genome-wide-testing DTC are related to: i)health, ii) curiosity, iii) ancestry, iv) contributing to research, and v) recreation. 6) Through a study of guidelines and population biobanks and companies selling genome-wide-testing we reported that there is a paucity of guidance on what to do with samples and data if a population biobank or DTC company closes (i.e.: is sold, or declares bankruptcy).[12] 7) By surveying clinical geneticists in Europe we found that more than 80% of clinicians are aware that DTC genetic testing exists and approximately half have seen at least one patient who had purchased a DTC genetic test. Furthermore, a majority of clinicians do not agree with the offer of genetic testing without face-to-face medical supervision, outside of the traditional health care system with no established patient-doctor relationship. This is especially true for serious medical conditions and for those that are neither treatable nor preventable. Finally the majority of respondents somewhat or strongly agree with the legal banning of genome-wide-testing as well as prenatal gender testing. 8) A study of the legal context showed that France, Germany, Portugal, and Switzerland have specific legislation that defines that genetic tests can only be carried out by a medical doctor after the provision of sufficient information concerning the nature, meaning and consequences of the genetic test and after the consent of the person concerned. Belgium and the United Kingdom allow the provision of DTC genetic tests. Although relevant legislation that bind DTC companies exists at the European level (E.g. the in vitro medical diagnostic devices, consumer protection legislation, data protection legislation), the lack of a harmonized (European) approach at all levels is problematic in a context where services are offered through the internet.

IMPACT: Through literature reviews, content analysis of company websites and surveys, we have brought to light, discussed at length and analysed the different ethical, legal and social issues associated with the existence of DTC genetic testing. Through the publication of our work in international peer reviewed journals as well as the organization of workshops and presentation at conferences, we have contributed to the education of different stakeholders (mostly lab researchers and health care professionals such as doctors, nurses and genetic counsellors) about these services, and have initiated and helped support a much-needed discussion on DTC genetic testing. Furthermore, our work has also been presented to policy makers as they attempt to decide on the best way to regulate these services. This work has also been helpful in the ultimate drafting of guidelines by the European Society of Human Genetics (2010). Along with the work of other researchers, it has contributed to the growing corpus of information that questions the wisdom of offering genetic testing outside of the traditional health care system for commercial purposes. Such information may have, in some way, contributed to the recent interest, in the last year, of the Food and Drug Association (USA) to increase the regulatory control over the sale of genetic tests DTC. Finally, the attention from the popular press regarding the study on clinicians' views will have helped educate the general public regarding genetic testing.

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